PRSS1 gene

protease, serine 1

Normal Function

The *PRSS1* gene provides instructions for making an enzyme called cationic trypsinogen. This enzyme is a serine peptidase, which is a type of enzyme that cuts (cleaves) other proteins into smaller pieces. Cationic trypsinogen is produced in the pancreas and helps with the digestion of food. Cationic trypsinogen is secreted by the pancreas and transported to the small intestine, where it is cleaved to form trypsinogen. When the enzyme is needed, trypsinogen is cleaved again into its working (active) form called trypsin. Trypsin aids in digestion by cutting protein chains at the protein building blocks (amino acids) arginine or lysine, which breaks down the protein. Trypsin also turns on (activates) other digestive enzymes that are produced in the pancreas to further facilitate digestion.

A particular region of trypsin is attached (bound) to a calcium molecule. As long as trypsin is bound to calcium, the enzyme is protected from being broken down. When digestion is complete and trypsin is no longer needed, the calcium molecule is removed from the enzyme, which allows trypsin to be broken down.

Health Conditions Related to Genetic Changes

hereditary pancreatitis

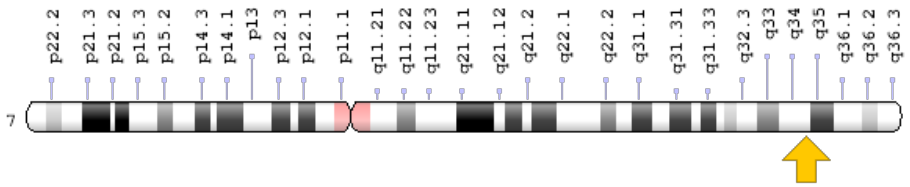
More than 40 mutations in the *PRSS1* gene have been found to cause hereditary pancreatitis, a condition characterized by recurrent episodes of inflammation of the pancreas (pancreatitis), which can lead to a loss of pancreatic function. Most of these mutations change single protein building blocks (amino acids) in cationic trypsinogen. Some *PRSS1* gene mutations result in the production of a cationic trypsinogen enzyme that is prematurely converted to trypsin while it is still in the pancreas. Other mutations prevent trypsin from being broken down. The most common *PRSS1* gene mutation that causes hereditary pancreatitis replaces the amino acid arginine with the amino acid histidine at position 122 in the enzyme (written Arg122His or R122H). As a result of this mutation, the enzyme is not able to be broken down, even when it is no longer bound to calcium.

Trypsin activity in the pancreas can damage pancreatic tissue and can also trigger an immune response, causing inflammation in the pancreas and leading to episodes of pancreatitis.

Chromosomal Location

Cytogenetic Location: 7q34, which is the long (q) arm of chromosome 7 at position 34

Molecular Location: base pairs 142,740,235 to 142,753,076 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- beta-trypsin
- cationic trypsinogen
- protease, serine, 1 (trypsin 1)
- TRP1
- TRY1
- TRY1_HUMAN
- TRY4
- TRYP1
- trypsin-1
- trypsin-1 preproprotein
- trypsinogen 1
- trypsinogen A

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Generation of Trypsin from Trypsinogen Leads to the Activation of Other Zymogens
  https://www.ncbi.nlm.nih.gov/books/NBK22589/#A1395

GeneReviews

- Pancreatitis Overview
  https://www.ncbi.nlm.nih.gov/books/NBK190101
- PRSS1-Related Hereditary Pancreatitis
  https://www.ncbi.nlm.nih.gov/books/NBK84399

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PRSS1%5BTIAB%5D%29+OR+%28cationic+trypsinogen%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- PROTEASE, SERINE, 1
  http://omim.org/entry/276000

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PRSS1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PRSS1%5Bgene%5D
- HGNC Gene Family: Proteases, serine
  http://www.genenames.org/cgi-bin/genefamilies/set/738
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9475
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5644
- UniProt
  http://www.uniprot.org/uniprot/P07477

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17003641
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